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TECH CENTER



1600

## RAW SEQUENCE LISTING

DATE: 07/25/2003

PATENT APPLICATION: US/09/109,082A

TIME: 14:44:57

#31

Input Set : A:\2003-07-17 2121-0140P.ST25.txt

Output Set: N:\CRF4\07252003\I109082A.raw

3 <110> APPLICANT: MELKI, Judith  
4 MUNNICH, Arnold  
6 <120> TITLE OF INVENTION: Spinal Muscular Atrophy Diagnostic Methods  
8 <130> FILE REFERENCE: 2121-0140P  
10 <140> CURRENT APPLICATION NUMBER: 09/109,082A  
11 <141> CURRENT FILING DATE: 1998-07-02  
13 <150> PRIOR APPLICATION NUMBER: 08/545,196  
14 <151> PRIOR FILING DATE: 1995-10-19  
16 <160> NUMBER OF SEQ ID NOS: 65  
18 <170> SOFTWARE: PatentIn version 3.2  
20 <210> SEQ ID NO: 1  
21 <211> LENGTH: 347  
22 <212> TYPE: DNA  
23 <213> ORGANISM: Homo sapiens  
25 <400> SEQUENCE: 1

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28	ccaggtctca	agtgatcccc	ctacctccgc	ctcccaaagt	tgtgggattg	taggcatgag	120
30	ccactgcaag	aaaaccttaa	ctgcagccta	ataattgttt	tctttgggat	aacttttaaa	180
32	gtacattaaa	agactatcaa	cttaatttct	gatcatat	tggtgaataa	aataagtaaa	240
34	atgtcttggtg	aacaaaatgc	tttttaacat	ccatataaag	ctatctatat	atagctatct	300
36	atgtctatat	agctattttt	tttaacttcc	ttttattttc	cttacag		347

39 <210> SEQ ID NO: 2  
40 <211> LENGTH: 444  
41 <212> TYPE: DNA  
42 <213> ORGANISM: Homo sapiens  
44 <400> SEQUENCE: 2

45	gtaagtctgc	cagcattatg	aaagtgaatc	ttacttttgt	aaaactttat	ggtttgtgga	60
47	aaacaaatgt	ttttgaacag	ttaaaaagtt	cagatgttaa	aaagttgaaa	ggttaatgta	120
49	aaacaatcaa	tattaaagaa	ttttgatgcc	aaaactatta	gataaaaggt	taatctacat	180
51	ccctactaga	attctcatat	ttaactgggt	ggttatgtgg	aagaaacata	ctttcacaat	240
53	aaagagcttt	aggatatgat	gccattttat	atcactagta	ggcagaccag	cagacttttt	300
55	tttattgtga	tatgggataa	cctaggcata	ctgcactgta	caactctgaca	tatgaagtgc	360
57	tctagtcaag	tttaactggg	gtccacagag	gacatgggtt	aactggaatt	cgtcaagcct	420
59	ctggttctaa	tttctcat	gcag				444

62 <210> SEQ ID NO: 3  
63 <211> LENGTH: 347  
64 <212> TYPE: DNA  
65 <213> ORGANISM: Homo sapiens  
67 <400> SEQUENCE: 3

68	aatttttaaa	ttttttgtag	agacagggtc	tcattatggt	gcccagggtg	gtgtcaagct	60
70	ccaggtctca	agtgatcccc	ctacctccgc	ctcccaaagt	tgtgggattg	taggcatgag	120
72	ccactgcaag	aaaaccttaa	ctgcagccta	ataattgttt	tctttgggat	aacttttaaa	180
74	gtacattaaa	agactatcaa	cttaatttct	gatcatat	tggtgaataa	aataagtaaa	240

p.6

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76 atgtcttgtg aacaaaatgc tttttaacat ccatataaag ctatctatat atagctatct      300
78 atatctatat agctattttt ttttaacttcc ttttattttc cttacag                    347
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83 <212> TYPE: DNA
84 <213> ORGANISM: Homo sapiens
86 <400> SEQUENCE: 4
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89 aaacaaatgt ttttgaacag ttaaaaagtt cagatgtagt aaagttgaaa ggtaaagtga     120
91 aaacaatcaa tattaagaa ttttgatgcc aaaactatta gataaaaggt taatctacat     180
93 ccctactaga atttcatac ttaactgggt gggtgtgtgg aagaaacata ctttcacaat     240
95 aaagagcttt aggatatgat gccattttat atcactagta ggcagaccag cagacttttt     300
97 tttattgtga tatgggataa cctaggcata ctgcactgta cactctgaca tatgaagtgc     360
99 tctagtcaag ttttaactgg gtccacagag gacatgggtt aactggaatt cgtcaagcct     420
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105 <211> LENGTH: 25
106 <212> TYPE: DNA
107 <213> ORGANISM: Artificial Sequence
109 <220> FEATURE:
110 <223> OTHER INFORMATION: R111 primer/probe characteristic of exon 8 of the T-BCD541
gene.
112 <400> SEQUENCE: 5
113 agactatcaa cttaattttct gatca                                           25
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117 <211> LENGTH: 24
118 <212> TYPE: DNA
119 <213> ORGANISM: Artificial Sequence
121 <220> FEATURE:
122 <223> OTHER INFORMATION: 541C770 primer/probe characteristic of exon 8 of the T-
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123 gene.
125 <400> SEQUENCE: 6
126 taaggaatgt gagcaccttc cttc                                           24
129 <210> SEQ ID NO: 7
130 <211> LENGTH: 23
131 <212> TYPE: DNA
132 <213> ORGANISM: Artificial Sequence
134 <220> FEATURE:
135 <223> OTHER INFORMATION: 541C960 primer/probe characteristic of exon 8 of the T-
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136 gene.
138 <400> SEQUENCE: 7
139 gtaataacca aatgcaatgt gaa                                           23
142 <210> SEQ ID NO: 8
143 <211> LENGTH: 20
144 <212> TYPE: DNA
145 <213> ORGANISM: Artificial Sequence
147 <220> FEATURE:
148 <223> OTHER INFORMATION: 541C1120 primer/probe characteristic of exon 8 of the T-
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149 gene.
151 <400> SEQUENCE: 8

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Input Set : A:\2003-07-17 2121-0140P.ST25.txt

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152 ctacaacacc cttctcacag 20  
 155 <210> SEQ ID NO: 9  
 156 <211> LENGTH: 294  
 157 <212> TYPE: PRT  
 158 <213> ORGANISM: Homo sapiens  
 160 <400> SEQUENCE: 9  
 162 Met Ala Met Ser Ser Gly Gly Ser Gly Gly Gly Val Pro Glu Gln Glu  
 163 1 5 10 15  
 166 Asp Ser Val Leu Phe Arg Arg Gly Thr Gly Gln Ser Asp Asp Ser Asp  
 167 20 25 30  
 170 Ile Trp Asp Asp Thr Ala Leu Ile Lys Ala Tyr Asp Lys Ala Val Ala  
 171 35 40 45  
 174 Ser Phe Lys His Ala Leu Lys Asn Gly Asp Ile Cys Glu Thr Ser Gly  
 175 50 55 60  
 178 Lys Pro Lys Thr Thr Pro Lys Arg Lys Pro Ala Lys Lys Asn Lys Ser  
 179 65 70 75 80  
 182 Gln Lys Lys Asn Thr Ala Ala Ser Leu Gln Gln Trp Lys Val Gly Asp  
 183 85 90 95  
 186 Lys Cys Ser Ala Ile Trp Ser Glu Asp Gly Cys Ile Tyr Pro Ala Thr  
 187 100 105 110  
 190 Ile Ala Ser Ile Asp Phe Lys Arg Glu Thr Cys Val Val Val Tyr Thr  
 191 115 120 125  
 194 Gly Tyr Gly Asn Arg Glu Glu Gln Asn Leu Ser Asp Leu Leu Ser Pro  
 195 130 135 140  
 198 Ile Cys Glu Val Ala Asn Asn Ile Glu Gln Asn Ala Gln Glu Asn Glu  
 199 145 150 155 160  
 202 Asn Glu Ser Gln Val Ser Thr Asp Glu Ser Glu Asn Ser Arg Ser Pro  
 203 165 170 175  
 206 Gly Asn Lys Ser Asp Asn Ile Lys Pro Lys Ser Ala Pro Trp Asn Ser  
 207 180 185 190  
 210 Phe Leu Pro Pro Pro Pro Pro Met Pro Gly Pro Arg Leu Gly Pro Gly  
 211 195 200 205  
 214 Lys Pro Gly Leu Lys Phe Asn Gly Pro Pro Pro Pro Pro Pro Pro Pro  
 215 210 215 220  
 218 Pro Pro His Leu Leu Ser Cys Trp Leu Pro Pro Phe Pro Ser Gly Pro  
 219 225 230 235 240  
 222 Pro Ile Ile Pro Pro Pro Pro Pro Ile Cys Pro Asp Ser Leu Asp Asp  
 223 245 250 255  
 226 Ala Asp Ala Leu Gly Ser Met Leu Ile Ser Trp Tyr Met Ser Gly Tyr  
 227 260 265 270  
 230 His Thr Gly Tyr Tyr Met Gly Phe Arg Gln Asn Gln Lys Glu Gly Arg  
 231 275 280 285  
 234 Cys Ser His Ser Leu Asn  
 235 290  
 238 <210> SEQ ID NO: 10  
 239 <211> LENGTH: 1582  
 240 <212> TYPE: DNA  
 241 <213> ORGANISM: Homo sapiens  
 243 <400> SEQUENCE: 10

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244 cggggcccca cgctgcgcac ccgcggggtt gctatggcga tgagcagcgg cggcagtggg      60
246 ggcggcggtcc cggagcagga ggattccgtg ctgttccggc gcggcacagg ccagagcgat      120
248 gattctgaca tttgggatga tacagcactg ataaaagcat atgataaagc tgtggcttca      180
250 tttaaagcatg ctctaaagaa tgggtgacatt tgtgaaactt cgggtaaacc aaaaaccaca      240
252 cctaaaagaa aacctgctaa gaagaataaa agccaaaaga agaatactgc agcttcctta      300
254 caacagtggg aagtgtggga caaatgttct gccatttggg cagaagacgg ttgcatttac      360
256 ccagctacca ttgcttcaat tgattttaag agagaaacct gtgttgtggg ttacactgga      420
258 tatggaaata gagaggagca aaatctgtcc gatctacttt cccaatctg tgaagtagct      480
260 aataatatag aacagatgc tcaagagaat gaaaatgaaa gccaagtttc aacagatgaa      540
262 agtgagaact ccaggtctcc tggaaataaa tcagataaca tcaagcccaa atctgctcca      600
264 tggaaaccct ttctccctcc accaccccc atgccagggc caagactggg accaggaaag      660
266 ccaggtctaa aattcaatgg cccaccaccg ccaccgccac caccaccacc ccacttacta      720
268 tcatgctggc tgcttccatt tccttctgga ccaccaataa ttccccacc acctcccata      780
270 tgtccagatt ctcttgatga tgctgatgct ttgggaagta tgtaatttc atggtacatg      840
272 agtggctatc atactggcta ttatatgggt tttagacaaa atcaaaaaga aggaagggtgc      900
274 tcacattcct taaattaagg agaatgctg gcatagagca gcactaaatg acaccactaa      960
276 agaaacgac agacagatct ggaatgtgaa gcgttataga agataactgg cctcatttct      1020
278 tcaaatatc aagtgttggg aaagaaaaaa ggaagtggaa tgggtaactc ttcttgatta      1080
280 aaagtatatg aataaccaaa tgcaatgtga aatattttac tggactcttt tgaaaaacca      1140
282 tctgtaaaag actgaggtgg ggggtgggag ccagcacggt ggtgaggcag ttgagaaaat      1200
284 ttgaatgtgg attagatttt gaatgatatt ggataattat tggtaatttt atggcctgtg      1260
286 agaagggtgt ttagtattat aaaagactgt cttaatttgc atacttaagc atttaggaat      1320
288 gaagtgttag agtgtcttaa aatgtttcaa atggtttaac aaaatgtatg tgaggcgtat      1380
290 gtggcaaaat gttacagaat ctaactggtg gacatggctg ttcattgtac tgtttttttc      1440
292 tatcttctat atgtttaaaa gtatataata aaaatattta attttttttt aaaaaaaaaa      1500
294 aaaaaaaaaa aaaaaaaaaa aaaaaaaaaa aaaaaaaaaa aaaaaaaaaa aaaaaaaaaa      1560
296 aaaaaaaaaa aaaaaaaaaa aa
299 <210> SEQ ID NO: 11
300 <211> LENGTH: 1408
301 <212> TYPE: DNA
302 <213> ORGANISM: Homo sapiens
304 <400> SEQUENCE: 11
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307 ccaggtctca agtgatcccc ctacctccgc ctcccaaagt tgtgggattg taggcagag      120
309 ccactgcaag aaaaccttaa ctgcagccta ataattgttt tctttgggat aactttttaa      180
311 gtacattaaa agactatcaa cttaatttct gatcatattt tgttgaataa aataagtaaa      240
313 atgtcttgtg aacaaaatgc tttttaacat ccatataaag ctatctatat atagctatct      300
315 atatctatat agctattttt ttttaacttc ttttattttc cttacagggt tttagacaaa      360
317 atcaaaaaga aggaagggtgc tcacattcct taaattaagg agtaagtctg ccagcattat      420
319 gaaagtgaat ctacttttg taaaacttta tggtttgtgg aaaacaaatg tttttgaaca      480
321 gttaaaaagt tcagatgtta gaaagttgaa aggttaatgt aaaacaatca atattaaaga      540
323 attttgatgc caaaactatt agataaaaag ttaacttaca tccctactag aattctcata      600
325 cttaatggtg tggttgtgtg gaagaaacat actttcaca taaagagctt taggatatta      660
327 tgccatttta tatcactagt aggcagacca gcagactttt ttttattgtg atatgggata      720
329 acctaggcat actgcactgt acactctgac atatgaagtg ctctagtcaa gtttaactgg      780
331 tgtccacaga ggacatggtt taactggaat tcgtcaagcc tctggttcta atttctcatt      840
333 tgcaggaaat gctggcatag agcagcacta aatgacacca ctaaagaaac gatcagacag      900
335 atctggaatg tgaagcgtta tagaagataa ctggcctcat ttcttcaaaa tatcaagtgt      960
337 tgggaaagaa aaaaggaagt ggaatgggta actcttcttg attaaaagtt atgtaataac      1020

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339 caaatgcaat gtgaaatatt ttactggact cttttgaaaa accatctgta aaagactgag 1080
341 gtgggggttg gaggccagca cgggtggtgag gcagttgaga aaatttgaat gtggattaga 1140
343 ttttgaatga tattggataa ttattggtaa ttttatggcc tgtgagaagg gtgttgtagt 1200
345 ttataaaaga ctgtcttaat ttgcatactt aagcatttag gaatgaagtg ttagagtgtc 1260
347 ttaaaatgtt tcaaatgggt taacaaaatg tatgtgaggc gtatgtggca aaatgttaca 1320
349 gaatctaact ggtggacatg gctgttcatt gtactgtttt tttctatctt ctatatgttt 1380
351 aaaagtatat aataaaaaata ttttaattt 1408

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354 &lt;210&gt; SEQ ID NO: 12

355 &lt;211&gt; LENGTH: 1582

356 &lt;212&gt; TYPE: DNA

357 &lt;213&gt; ORGANISM: Homo sapiens

359 &lt;400&gt; SEQUENCE: 12

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360 cggggcccca cgctgcgcat ccgcgggttt gctatggcga tgagcagcgg cggcagtggt 60
362 ggcggcgtcc cggagcagga ggattccgtg ctgttccggc gcggcacagg ccagagcgat 120
364 gattctgaca tttgggatga tacagcactg ataaaaagcat atgataaagc tgtggcttca 180
366 ttttaagcatg ctctaaagaa tgggtgacatt tgtgaaactt cgggtaaacc aaaaaccaca 240
368 cctaaaagaa aacctgctaa gaagaataaa agccaaaaga agaatactgc agcttcctta 300
370 caacagtgga aagttgggga caaatgttct gccatttggg cagaagacgg ttgcatttac 360
372 ccagctacca ttgcttcaat tgattttaag agagaaacct gtgttggtgt ttacactgga 420
374 tatggaaata gagaggagca aaatctgtcc gatctacttt ccccaatctg tgaagtagct 480
376 aataatatag aacagaatgc tcaagagaat gaaaatgaaa gccaaagttc aacagatgaa 540
378 agtgagaact ccagggtctcc tggaaataaa tcagataaca tcaagcccaa atctgctcca 600
380 tggaaactctt ttctccctcc accacccccc atgccagggc caagactggg accaggaaag 660
382 ccaggtctaa aattcaatgg cccaccaccg ccaccgccac caccaccacc ccacttacta 720
384 tcatgctggc tgcctccatt tccttctgga ccaccaataa ttccccacc acctcccata 780
386 tgtccagatt ctcttgatga tgctgatgct ttgggaagta tgttaatttc atggtacatg 840
388 agtggctatc atactggcta ttatatgggt ttcagacaaa atcaaaaaga aggaagggtc 900
390 tcacattcct taaattaagg agaaatgctg gcatagagca gcactaaatg acaccactaa 960
392 agaaacgatc agacagatct ggaatgtgaa gcgttataga agataactgg cctcatttct 1020
394 tcaaaatatc aagtgttggg aaagaaaaaa ggaagtggaa tgggtaactc ttcttgatta 1080
396 aaagtatatg aataaccaa tgcaatgtga aatattttac tggactctt tgaaaaacca 1140
398 tctgtaaaag actggggtgg ggggtggagg ccagcacggt ggtgaggcag ttgagaaaat 1200
400 ttgaatgtgg attagatttt gaatgatatt ggataattat tggtaatttt atggcctgtg 1260
402 agaagggtgt tgtagtttat aaaagactgt ctttaatttg atacttaagc atttaggaat 1320
404 gaagtgttag agtgtcttaa aatgtttcaa atgggttaac aaaatgtatg tgaggcgtat 1380
406 gtggcaaaat gttacagaat ctaactggtg gacatggctg ttcattgtac tgttttttc 1440
408 tatcttctat atgtttaaaa gtatataata aaaatattta atttttttt aaaaaaaaaa 1500
410 aaaaaaaaaa aaaaaaaaaa aaaaaaaaaa aaaaaaaaaa aaaaaaaaaa aaaaaaaaaa 1560
412 aaaaaaaaaa aaaaaaaaaa aa 1582

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416 &lt;211&gt; LENGTH: 1408

417 &lt;212&gt; TYPE: DNA

418 &lt;213&gt; ORGANISM: Homo sapiens

420 &lt;400&gt; SEQUENCE: 13

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423 ccagggtctca agtgatcccc ctacctccgc ctcccaaagt tgtgggattg taggcatgag 120
425 ccactgcaag aaaaccttaa ctgcagccta ataattgttt tctttgggat aactttttaa 180
427 gtacattaaa agactatcaa cttaatttct gatcatattt tgttgaataa aataagtaaa 240
429 atgtcttggt aacaaaatgc tttttaacat ccatataaag ctatctatat atagctatct 300

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RAW SEQUENCE LISTING ERROR SUMMARY  
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Please Note:

Use of n and/or Xaa have been detected in the Sequence Listing. Please review the Sequence Listing to ensure that a corresponding explanation is presented in the <220> to <223> fields of each sequence which presents at least one n or Xaa.

Seq#:14; N Pos. 7,14,43,84,204,244,250,316,319,332,345,348  
Seq#:15; N Pos. 11,44,66,67,94,108  
Seq#:18; N Pos. 26,69,72,73,88,120,158,277,298  
Seq#:22; N Pos. 330,450